

Claims:

1. An isolated nucleotide sequence according to SEQ ID NO:3, a functional fragment thereof, or a sequence that hybridizes thereto.

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2. An isolated polypeptide sequence according to SEQ ID NO:4 or a functional fragment thereof.

3. The use of at least one functional fragment of a nucleotide according to SEQ ID NO:1 or a peptide according to SEQ ID NO:2 for preparing a pharmaceutical composition for the treatment of a gene disorder marked by the presence of a mutation at a position corresponding to position 298 of SEQ ID NO:3.

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4. An isolated nucleic acid molecule according to Claim 1 in the form of a plasmid.

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5. A vector comprising the nucleic acid according to Claim 1 or a nucleic acid encoding the polypeptide of Claim 2.

6. A vector according to Claim 5, wherein the vector is a virus, such as a DNA virus or a retrovirus.

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7. A vector according to Claim 6, wherein the vector is selected from the group consisting of adeno-associated virus, adenovirus, herpesvirus, MoMLV, HIV-1, and SIV.

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8. A host cell transformed or transfected with a vector according to any one of Claims 5-7, such as a eukaryotic cell, a COS cell, a prokaryotic cell, a 293EBNA cell, or an insect cell.

9. A host cell transformed or transfected with a vector comprising a nucleotide sequence according to Claim 1, operatively linked to a promoter, such that said host cell expresses a mutated NGFB protein.

5 10. A molecular probe for the indication a genetic defect, comprising:  
a nucleotide sequence according to SEQ ID NO:3 or a sequence which hybridises to said nucleotide sequence under stringent conditions; and  
a label for detecting the presence of said sequence, such as a radioactive label.

10 11. A method of screening a patient for a genetic defect, comprising:  
obtaining a sample of genetic material from said patient, and  
identifying the nucleotide present at a position corresponding to position 298 of  
SEQ ID NO:3,  
wherein said patient has a genetic defect if a nucleotide other than cytosine is  
15 identified.

12. A method for detecting the presence of a genetic defect in a biological sample,  
comprising:  
contacting the biological sample with a nucleic acid molecule comprising a  
20 compliment to SEQ ID NO:3 as a probe in a nucleic acid hybridization assay; and  
detecting whether the nucleic acid molecule has undergone hybridization,  
wherein hybridization indicates the presence of a genetic defect in the biological  
sample.

25 13. A transgenic animal comprising a modified nucleotide at a position  
corresponding to position 298 of SEQ ID NO:3, such as a thymine.

14. A transgenic animal according to Claim 13, wherein the animal is a mammal,  
such as a rodent.

15. A transgenic animal comprising one or more cells which express a sequence according to SEQ ID NO:3.

16. A method of evaluating the ability of a potential therapy to treat or cure a genetic disorder, comprising:
- 5 administering the potential therapy to a transgenic animal according to any one of Claims 13-14; and
- evaluating a pain response in said animal,
- wherein an improved pain response in said animal as compared to untreated
- 10 similarly-situated transgenic animals indicates that the potential therapy is able to treat or cure a genetic disorder.